

Rinophyma in ASMD (Niemann-Pick Disease Type B)

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Clinical Image

Recognizing the facies in specific diseases has always been considered a valuable aid in medicine for the diagnosis. Here we show a peculiar skin involvement, as rinophyma in a man of 44-year old, affected by Niemann-Pick disease type B. Diagnosis was established when he was 1-years-old in the Bambino Gesù Childrens Hospital, because of hepatosplenomegaly. The skin appeared thickened and oily in infancy; the face and nose gradually changed until they reached their present appearance.

Niemann-Pick disease type B (NP-B), known as the 'visceral' form of acid sphingomyelinase (ASMD) deficiency, is caused by homozygous or compound heterozygous mutations in the sphingomyelin phosphodiesterase-1 gene (SMPD1; 607608), which encodes acid sphingomyelinase (ASM) on chromosome 11p15. Differently from the progressive and fatal infantile neurovisceral form (NP-A), NP-B patients may survive into adulthood without central nervous system signs. Serum triglycerides and LDL-cholesterol are often elevated, while HDL-cholesterol is low. The primary organ systems affected in all ASMD patients are the spleen, the liver and the lung, with accumulation of sphingomyelin and other lipids in the cells of the reticuloendothelial system. Indeed, lipid filled foam cells that can be readily detected also in the skin.

Hopefully the Olipudase alfa, the first specific treatment for ASMD, will provide sustained improvements in clinical feature in these patients [1].



Rhinophyma (a name derived from greek "rhis," meaning nose, and "phyma," for skin tumor) is a nasal deformity due to the proliferation of sebaceous glands and underlying connective tissue.

It is most commonly found in white men with a male to female ratio of 5 to 1 to 30 to 1, supporting the hypothesis that androgens enhance rhinophyma development [4].

Skin changes in ASMD did not often receive enough attention. Indeed, cultured skin fibroblasts from affected patients are less active to degrade sphingomyelin to ceramide and phosphocholine in lysosomes [2]. Sphingomyelin storage was present in multiple cell types, including dermal fibroblasts, macrophages, vascular endothelial cells, vascular smooth muscle cells, perineurium, and Schwann cells as confirmed by electron microscopy [3].

Histopathologic examination reveals hypertrophic sebaceous glands and thickened dermis containing fibrovascular myxoid stroma and lymphatic cells. Expansion of the sebaceous unit leads to sebum plugging. In observed cases of severe rhinophyma, sebaceous glands become destroyed by edema and fibrosis, producing a lymphoedema-type histologic picture [3].

Etiologies and Differential Diagnosis

Usually, rhinophyma is considered the final and most severe stage of acne rosacea, primarily affecting white males over the age of 50.

It can be mimicked by several conditions: adenoid squamous cell carcinoma, squamous cell carcinoma, sebaceous adenoma, sebaceous carcinoma, and angiosarcoma have also been histologically diagnosed within rhinophyma. Sarcoidosis, lymphoma, metastatic lung cancer, and granuloma eosinophilicum have all posed clinical similarities to rhinophyma [5].

Basal cell carcinoma is suggested to develop in 3 to 10% of rhinophyma patients and can easily hide within the deformed nodular skin Keefe et al. reported an assumed rhinophyma later pathologically diagnosed as entirely composed of basal cell carcinoma [6].

This case shows the Rynophyma in type B ASMD (Niemann-Pick disease) too.

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